Celiac Disease Genomic, Environmental, Microbiome and Metabolomic Study

55 Fruit Street (Jackson 14) • Attn: CDGEMM Study • Boston, MA 02114

The CDGEMM Study enrolls infants who have a parent or sibling diagnosed with celiac disease (CD). The risk of developing CD for these infants is increased by 8-25% over that of the general population. Enrolled children are followed from birth until they reach five years of age, including periodic monitoring for signs of CD. A main aim of the study is to track the development of the gut microbiome by collecting the child's stool samples and watching how the microbial communities evolve over time. We hope to identify a distinct microbial pattern that will allow us to predict who will develop CD before it happens so that we can learn how to prevent it.

CDGEMM by the Numbers

Since the kick-off of CDGEMM, we have:

- ☑ Enrolled **90 children** in **25 of the 50** United States
- ☑ Enrolled **75 children** in Italy
- ☑ Collected **400** poop samples
- ☑ Conducted **155** celiac antibody tests

Determining each GEMM's risk for CD

One of the benefits of the CDGEMM Study includes close monitoring of each GEMM for celiac disease. Each time blood is drawn it is tested for the antibodies that are used to support the diagnosis of celiac disease. This testing allows for early detection and treatment if necessary. In addition, genetic testing is performed to assess whether the child has the genes compatible with celiac disease.

For babies enrolled at birth, there is the option to provide a small sample of cord blood, for which a portion can be used to perform the genetic testing. *For babies enrolled after birth*, a small amount of extra blood is collected at the 12 month study visit to perform the genetic testing.

Are you a study participant who recently received your child's genetic testing results? Here is a breakdown of what each result means:

- ◊ ***DQ2 (2 copies)*** – this means that your child carries two copies of the DQ2 gene (one from mother, one from father). This genetic type is considered the highest risk for developing celiac disease.
- ◊ ***DQ2*** – this means that your child only carries one copy (either from mother or father) of the DQ2 gene. This genetic type is compatible with celiac disease.
- ◊ ***DQ8*** – This means that your child only carries one copy (either from mother or father) of the DQ8 gene. This genetic type is considered lowest risk for developing celiac disease.
- ◊ ***DQ2/DQ8*** – this means that your child carries one copy of the DQ2 gene and one copy of the DQ8 gene. This genetic type is compatible with celiac disease.
- ◊ If your child's result returns as ***HLA negative*** (carrying neither the DQ2 nor the DQ8 gene), this means that he or she will never develop celiac disease.

While infants with a family history of CD have an 8-25% higher risk of developing CD compared to the general population, the majority of infants at risk for CD (even those with the highest risk) will not develop CD. Thus, there is currently no recommendation to refrain from gluten introduction in children with a high risk of developing celiac disease. Read more about introducing gluten to your baby here.

Click here to learn more about genetic testing for celiac disease, or email the CDGEMM Study team at CDGEMM@mgh.harvard.edu to discuss your child's results.

What's New with CDGEMM?

We're excited to introduce our 'Welcome Packet' which will soon be distributed to each GEMM at enrollment! This packet includes:

- ◊ A letter welcoming families to the study;
- ◊ Short biographies about the clinical staff members working on CDGEMM;
- ◊ An article describing how CDGEMM will make history for celiac disease;
- ◊ A study calendar to help keep track of which samples need to be collected and when; and
- ◊ A progress certificate with stickers to track each year that a GEMM completes!

Our study team has also created a Facebook group that is exclusively available to parents of participants in the study. This Facebook group was designed as a safe space to privately chat, ask questions, share pictures, and give positive words of encouragement to other families who are also raising a GEMM. Group participation is completely voluntary!

If you are currently enrolled in CDGEMM, request to join today by emailing us at CDGEMM@mgh.harvard.edu

Stay connected with CDGEMM!

Who's Talking about CDGEMM?

Allergic Living published an article in July 2016 about the high hopes that the CDGEMM study holds for future prevention of celiac disease. The article features Dr. Alessio Fasano, the principal investigator of the CDGEMM study, who emphasizes just how important this study is in understanding how the environment, genetics and the composition of microbial communities in our gut may influence the development of celiac disease.

Several online bloggers and websites recently helped spread the word about CDGEMM through postings on their social media accounts. *Gluten Free Living* published an article on their website and Facebook page, while one of our very own GEMM families generously agreed to be featured in an article published by *Beyond Celiac,* outlining their first-hand perspective of what participating in the study entails. Our study co-investigator, Dr. Maureen Leonard, was also featured on the *Celiac Project Podcast*, during which she spoke about how CDGEMM aims at uncovering the unknown triggers that, in combination with specific genetics and gluten ingestion, results in some individuals developing celiac disease at age 3 and others at age 53.

These, and several other exciting articles have been posted about the CDGEMM study and we are excited to continue to spread the word about CDGEMM across all social media platforms in the new year!

Help us reach 100 US GEMM's

We are still recruiting precious GEMMs for the CDGEMM Study. Children aged 6 months or younger who have a parent or sibling diagnosed with celiac disease are eligible to participate. **Consider sharing information about CDGEMM with your friends, relatives and/or patients with celiac disease!** Also, be sure to visit **www.CDGEMM.org** for more information about the study.